

# Ehlers-Danlos syndrome

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## ABSTRACT

Ehlers-Danlos syndrome (EDS) is a generalized disorder of one element of connective tissue manifesting clinically by fragility and hyperelasticity of the skin and joint laxity. It is a hereditary disorder, the inheritance being usually autosomal dominant with low penetrance. Autosomal recessive and X-linked recessive varieties are also known. First described by Hippocrates in 4<sup>th</sup> century B.C., the various clinical types with variable penetrance have been described lately. The number of cases EDS reported in the literature is very meagre. With the available information only about six publications of classic EDS in siblings had been reported in Indian literature.

**Key words:** Cigarette paper scarring, Gorlin's sign, Reverse Namaskar sign

## INTRODUCTION

Ehlers-Danlos syndrome (EDS) is a hereditary disorder of connective tissue characterized by fragility of skin and blood vessels, hyperextensibility of the skin and joint laxity. The diagnosis is mainly clinical. Milder variants of the classical mitis form of the EDS are common in the population and can be identified by a well-defined clinical scoring system.

## CASE REPORT

A 17-year-old female born out of nonconsanguineous marriage attended the dermatology out-patient department with a history of laxity of skin and joints. A history of bruising of the skin and gaping wounds with even very trivial trauma could be easily ascertained, but no episode of a major hemorrhagic accident had ever occurred in the past. The patient was an outcome of a full term normal delivery with no neonatal or perinatal morbidity. The family history revealed no such complaints in parents and siblings.

On physical examination, the skin was soft, velvety and hyperextensible [Figure 3] but with normal recoil. Atrophic cigarette paper scars [Figure 1] were seen on forehead, cheeks, forearms, elbows, knees and both shin. Joints were hypermobile [Figure 2] with genu recurvatum of the knee and hallux valgus deformity of both great toes. Gorlin's sign and reverse Namaskar sign [Figure 4] could be elicited. There was no evidence of ecchymosis, cyanosis, digital

clubbing, jaundice and lymphadenopathy. The fundus, routine blood test, urine, coagulogram, blood urea, sugar and echocardiogram were within the normal limits. X-ray of the spine and knee joints was normal except for feet which showed hallux valgus deformity. Histopathology with special stain for collagen and elastic fibers showed a decrease in collagen fibers with the relative increase in elastic fibers consistent with the diagnosis of EDS [Figures 5 and 6].

EDS cases have been reported in previous literature.<sup>[1-3,6,7]</sup> It's a heterogeneous group of inherited disorder of connective tissue manifesting clinically by fragility and hyperelasticity of the skin and joint laxity. It's a rare genetic disorder affecting 1:5000. The inheritance being usually autosomal dominant with low penetrance. Autosomal recessive and X-linked recessive varieties are also known. Van Meekeren<sup>[1]</sup> described hyperelastic skin and Koop described hypermobility of joints. Ehler noticed the easy bruisability of the skin whereas; Danlos noted the peculiar cigarette paper scars and pseudotumour formation of the skin. Danlos also put forth four diagnostic criteria, namely, hyperelasticity of skin [Figure 3], fragility of skin, hypermobility of joints [Figure 4] and subcutaneous molluscus pseudotumour formation. More than ten clinical types have been described based on clinical, genetic and biochemical information.

In a recent consensus in Villefranche, in 1997, the classification of EDS was reorganized into

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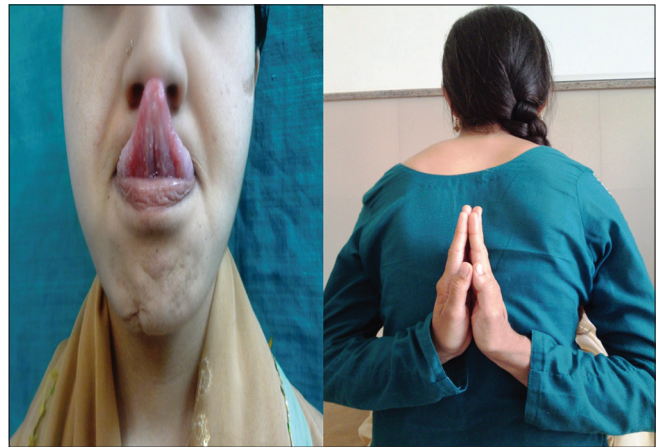
**Figure 1:** Cigarette paper scarring on forehead and lower limbs



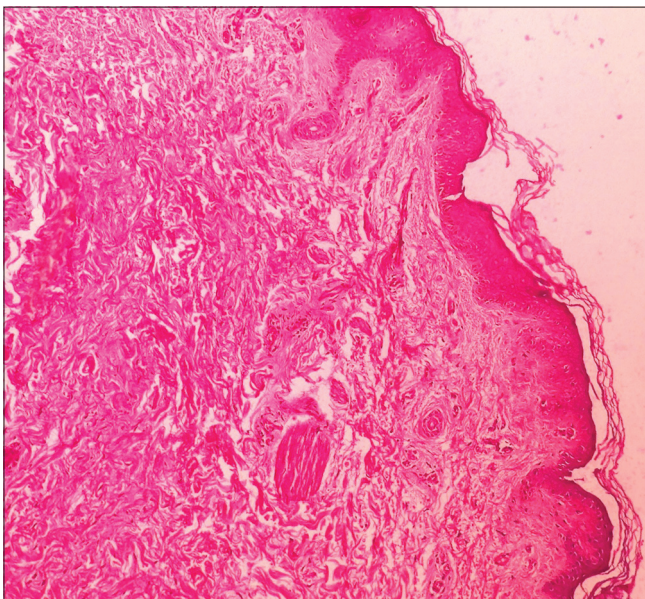
**Figure 2:** Hypermobile joints



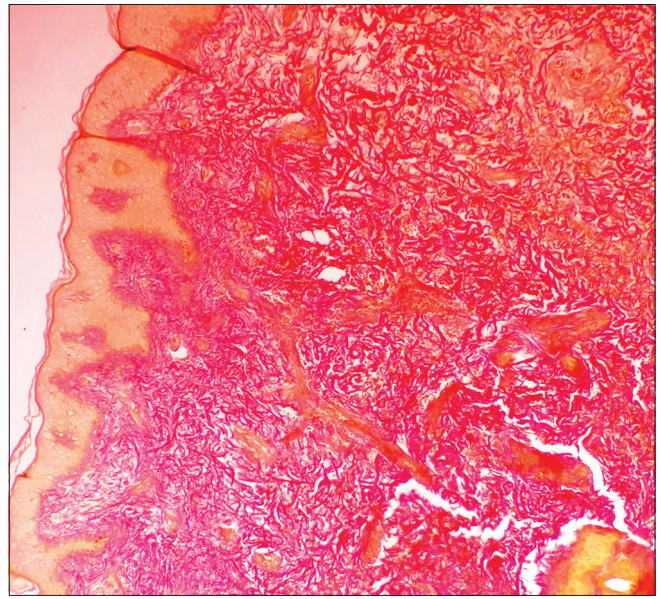
**Figure 3:** Hyperelastic skin and palms touching the floor



**Figure 4:** Gorlin's sign and Reverse Namaskar sign



**Figure 5:** Van Giesons stain for collagen fibers under ×10 showing decrease in collagen bundles



**Figure 6:** Acid Orcein Giemsa stain for elastic fibres under ×10 showing relative increase in elastic fibres

six major subtypes. The diagnostic skin signs described in EDS includes Gorlins sign<sup>[4]</sup> [Figure 4](ability to touch

the tip of the nose with the tongue), Metenier sign<sup>[5]</sup> (easy eversion of upper eyelid) and atrophic “cigarette paper”



scarring [Figure 1]. Molluscoid pseudotumors and spheroids may occur in EDS. These are subcutaneous nodules due to herniation of subcutaneous fatty tissue and resemble lipomas histologically. The “reverse Namaskar sign”<sup>[6]</sup> [Figure 4], a valuable diagnostic sign has been described by Premalatha in patients with EDS. Absence of ocular lesions, ecchymosis, large joint dislocations and periodontitis led us to classify our patient as EDS Type II (mitis). Treatment is highly unsatisfactory. Patients with EDS VI respond to oral ascorbic acid. We report this case because of the classical clinical signs present, which are depicted in the figures which will be helpful to diagnose the case of EDS in clinical practice.

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